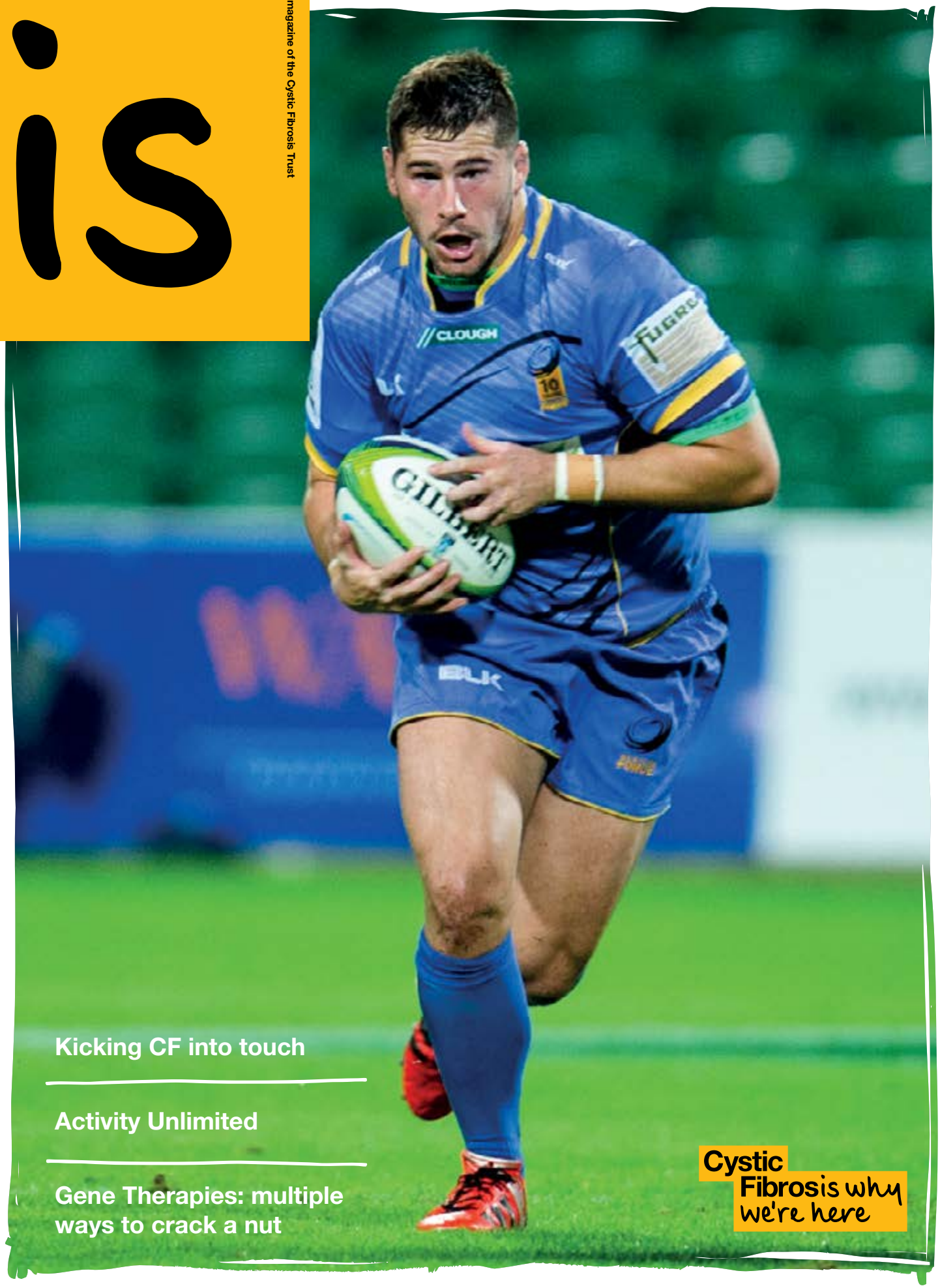
Issue 5: Autumn 2015

is

The magazine of the Cystic Fibrosis Trust



Kicking CF into touch

Activity Unlimited

Gene Therapies: multiple ways to crack a nut

Cystic Fibrosis why we're here

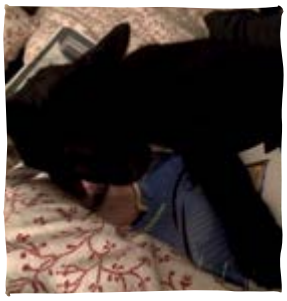
is

Regulars

News

Features

Welcome



Welcome to the latest issue of is! We're following a slight theme with this one: from cover star Nathan Charles (who you can meet on p 14) to tai chi research (see p11) and epic bike rides (see p 8), we're focusing on the importance of being active in managing cystic fibrosis. That's the driving force behind Activity Unlimited, a great new initiative the Trust is launching in 2016 (p 24).

Elsewhere we explore the exciting world of genetic therapies (p 20), and celebrate the amazing contribution from schools in raising funds and awareness (p 27). On p 28 you'll hear from a parent and a nurse about the emotional moment when a family first encounters cystic fibrosis, and how we are there to offer support and information.

Running through all of this is you, our supporters. Without your continued and generous support, none of the research and campaigns we invest in or the services we offer are possible. Thank you for everything you do – together we can make a life unlimited a reality for everyone affected by cystic fibrosis. Until next time, enjoy the Rugby World Cup and happy reading!

Henry Fogarty,
Editor

Trust Chairman George Jenkins OBE rides out with Jonathan King, who used exercise to get himself off the transplant list. 24

All communications should be sent to **Editor, is magazine, Cystic Fibrosis Trust, 2nd Floor, One Aldgate, London EC3N 1RE**

ismagazine@cysticfibrosis.org.uk

is magazine is published by the Cystic Fibrosis Trust registered as a charity in England and Wales (1079049) and in Scotland (SC040196). A company limited by guarantee registered in England and Wales number 3880213. Registered office 2nd Floor, One Aldgate, London EC3N 1RE.

Cover photo: Nathan Charles (see page 14).

Some pictures in this publication may be posted by models or taken from library images.

ISSN 2052-8337

Medical information included in is magazine is not intended to replace any advice you may receive from your doctor or CF multidisciplinary team and it is important that you seek medical advice whenever considering a change of treatment. Opinions expressed in articles do not necessarily express the official policy of the Cystic Fibrosis Trust. Information correct at time of going to press.

If you no longer want to receive is magazine or need to update your details, please contact the Trust's Supporter Care team (020 3795 2177; supportercare@cysticfibrosis.org.uk).

FRSB give with confidence

Share your thoughts on is magazine

- facebook.com/cftrust
- tweet @cftrust
- forum.cysticfibrosis.org.uk

Still in love with Bianca: Longtime Trust supporter Bianca Nicholas flew the flag for the UK and the CF community when she took part in the 2015 Eurovision Song contest, as one half of Electro Velvet.

News in brief

Aldgate

The Cystic FibrosisTrust has moved to central London as part of our wider plans to ensure we're in the best possible shape to transform the lives of people with cystic fibrosis.

Moving to our new home in Aldgate, and the technology it supports, means we are better integrated with the cystic fibrosis community.

Our new address is:
Cystic Fibrosis Trust,
One Aldgate, London EC3N 1RE.

NovaBiotics presents "potential breakthrough" drug

The first drug specifically targeting the infectious outbreaks associated with cystic fibrosis could be a step closer after NovaBiotics presented data from its Lynovex® study, co-funded by the Trust, at the European Cystic Fibrosis Conference in Brussels.

The Phase IIa clinical trial found that subjects could absorb and process cysteamine, brand name Lynovex®, in concentrations shown to minimise the impact of outbreaks of lung infection (exacerbations) for people with cystic fibrosis, which can cause regular hospital stays.

The Trust's Director of Strategic Innovation, Dr Janet Allen, said: "The Trust is delighted to see this promising data presented at the ECFS meeting. Part of our research strategy, launched in 2013, was to engage with the biotech sector - we are therefore pleased to see this set of data coming through."

Dr Deborah O'Neil, Chief Executive of NovaBiotics, said: "This encouraging data is a key step in the development of Lynovex® as a much needed therapy in CF exacerbations. For me, this data goes further to support the potential breakthrough that Lynovex® may offer in cystic fibrosis."

The next phase of clinical development is likely to commence within the next 12 months.

Don't forget you can find highlights of the latest news on our Twitter feed. Stay connected with the Trust at twitter.com/cftrust. **Follow us on Twitter @cftrust.**

#CFyelfie

Thank you to all who showed their support for this year's Wear Yellow Day by donating, posting photos on Facebook and tweeting photos on Twitter with the hashtag #CFyelfie.

Northern Irish CF community benefits from new scheme

The Trust has partnered with the Belfast Health and Social Care Trust Charitable Trust Funds and the Citizens Advice Bureau (CAB) to launch a year-long pilot scheme offering dedicated benefits advice for the CF community in Northern Ireland.

Launching the CAB initiative at the Trust's Regional Meeting in Belfast (left to right): Anne-Marie McGrade, Margaret McCafferty, Anne Calvert, Linda Williamson and Pat Colton.

The scheme provides two specially-trained CAB advisers offering advice on complicated benefits issues via phone, Skype and face-to-face meetings.

Mother-of-two Lorraine Mullen, who has CF, said the service would help with her changing circumstances: "The medical team are a huge help but they don't know about the benefits you are eligible for. "Knowing I have somewhere dedicated to helping me really helps to take away the stress and worry."

The Cystic Fibrosis Trust's Head of Information and Support, Jacqueline Ali, said the Trust was delighted to support the initiative given the cross-infection risks associated with cystic fibrosis: "Technologies such as Skype which provide 'face to face' interaction without the need to meet in person are an important way of delivering support to the cystic fibrosis community and reducing feelings of isolation."

The confidential service is available Monday 2–3pm, for benefits enquiries and to make appointments with an advisor, and Thursday 1–4pm, for appointments with an advisor.

Welcome to the Square: During CF Week BBC 1's 'Eastenders' introduced Jade, a new character with cystic fibrosis. Members of the Trust worked with producers on the depiction of the condition.

News in brief

Telephone Appeal

In the coming months we will be speaking to some of our supporters for our 2015 telephone appeal.

The money we raise will help support our vital research projects, such as our Strategic Research Centres (see p10), and your support is invaluable.

During last year's campaign over 700 supporters pledged £52,000 per year, and we would like to thank everyone involved.

For a fourth year our appeal partner is Ethicall, a specialist telephone fundraising agency.

If you have any questions about the appeal, please visit cysticfibrosis.org.uk/telephoneappeal, or call our supporter care team on **020 3795 2177**.

On the line: Members of the Ethicall team helping to raise vital funds to support the Trust.

Time to stop the clock on cystic fibrosis

The Cystic Fibrosis Trust is calling for greater access to precision medicines for people with cystic fibrosis, in a new campaign called 'Stop the Clock'.

As part of our fight for a life unlimited for people with cystic fibrosis, we are striving to make precision (or 'personalised') medicines available to over 90% of the UK's CF population within a decade.

Precision medicines tackle cystic fibrosis at its source, the defective CFTR channel, and target specific genotypes.

Only a little more than four per cent of people with cystic fibrosis in the UK currently have access to personalised medicines. These patients receive a drug called ivacaftor (Kalydeco) which has been shown to increase lung function, slow the progression of lung disease and more than halve the number of hospital admissions.

Later this year the use of ivacaftor could be extended to cover younger patients (age three to five years) and an additional genotype. A new combination drug, Orkambi, will also be appraised by the NHS; which could benefit around half the cystic fibrosis population, those aged 12 and older with two copies of the F508del genotype. There are several other promising treatments in the pipeline.

The Trust is committed to galvanising the cystic fibrosis community and supporters to support the fight for personalised medicines.

James Barrow, Head of External Affairs, said: "New drugs move us closer to a world in which people with cystic fibrosis can lead a life unlimited by the condition. However, access to personalised medicines is not guaranteed. We will fight to make sure that people can benefit from new treatments. Your help and views will play a crucial part in our campaign as we need to articulate a better future for people living with the condition, their families and carers."

Gene therapy trial results released

Professor Eric Alton.

Results of a clinical trial conducted by the Gene Therapy Consortium (GTC) and published in 'The Lancet Respiratory Medicine' demonstrate the potential for gene therapy to slow the decline of lung function in people with cystic fibrosis.

The GTC began the Multidose Gene Therapy Clinical (Wave 1) Trial in 2012 and the results suggest gene therapy could one day be added to the range of treatments used to manage cystic fibrosis.

Professor Eric Alton from the GTC said the participants received "modest but significant benefits," with 62 patients reporting an average 3.7% increase in FEV1 lung function compared to the control group, though the effect of the treatment was inconsistent.

The Trust's Chief Executive Ed Owen welcomed the results, which follow the Trust's announcement of £500,000 further funding for the GTC over the next two years:

"We will continue to invest in innovative genetic research and work with academic and industry partners to help us achieve our goal of a life unlimited by cystic fibrosis."

The Trust has invested £40 million in gene therapy since 2002 and continues to fund research into genetic therapies.

Find out more at cysticfibrosis.org.uk/gtcpresentation

End of the cycle: Peter van Heyningen raised more than £300,000 over his 30 years of London to Brighton bike rides.

Peter's Pedal Power

Few people know the 54-mile London to Brighton Bike Ride route as well as Peter van Heyningen, who completed his 30th (and final) London to Brighton in June.

Peter began fundraising for a different charity, but after his son Leo (now 26) was born with cystic fibrosis he started raising money for the Royal Brompton Hospital to fund a cystic fibrosis research registrar.

"I've always enjoyed cycling and originally took part in the bike ride to keep fit, but I'm 60 now and all good things must come to an end," Peter said.

"I received 90 donations from my friends and family and they've been extremely generous over the years, but I'm not sure I can ask them for any more sponsorship."

Peter and his teams have raised over £300,000 over the 30 years and he hopes that his team, Pedro's Pedallers, will continue riding and fundraising for the Trust for many years to come.
Speaking after his last ride, Peter said: "The conditions were perfect for riding and all nine of us made it in good time, including Professor Jane Davies who was one of the first registrars we funded, many years ago."

Don't forget you can find highlights of the latest news and read comments on our Facebook page. 'Like' our page and stay connected with the Trust at facebook.com/cftrust.

Championing change with Champneys

UK health spa chain Champneys launched its Charity of the Year partnership with the Trust at a family cycling day in May at the Henlow Grange resort in Bedfordshire, raising £15,000.

Champneys and the Trust hope to raise over £100,000 through the scheme, which runs until March 2016 and includes employee fundraising, events and sporting challenges as well as direct benefits for the CF community.

"As the parent of a child with cystic fibrosis, I have come to realise and appreciate the impact that employer engagement from within the wider CF community can have on raising awareness and funds to help beat cystic fibrosis for good," Champneys Group General Manager, Will Simmonds said.

"I would encourage anyone affected by the condition to engage with their employers in aid of the fight against cystic fibrosis."

From 1 September, anyone with cystic fibrosis in the UK or parents or carers of a child under 18 with the condition can have a fantastic two-night stay at any of Champneys' four resorts including accommodation, all meals, exercise classes and a treatment for nearly 50% off.

Visit cysticfibrosis.org.uk/champneys for full details.

Family fun: TV personality Sian Lloyd joins participants in charity partner Champneys' family cycling day.

Campaigning for change

1. Increase the number of successful lung transplants for people with cystic fibrosis.
2. Increase NHS capacity to appraise drugs for rare conditions.
3. Streamline European licensing and UK appraisal systems.
4. Urge industry and Government to work together to ensure equal access to new medicines.
5. Ensure the government invests now to beat cystic fibrosis.
6. Invest in specialist CF centres.
7. Protect standards of care.
8. Press for change on prescription charges.
9. Ensure Personal Independent Payment (PIP) works for people with cystic fibrosis.
10. Fight for our community and enhance wider understanding of cystic fibrosis in all walks of life.

The (CF) nation decided: Thank you to everyone who helped complete the Trust's manifesto, with a call for better education and awareness.

New research centres announced

In February the Trust announced two new £750,000 Strategic Research Centres (SRC), virtual centres of excellence, bringing together the brightest and best scientists from across the UK and beyond to tackle specific problems within the field of cystic fibrosis.

'F508del-CFTR SRC', led by Dr David Sheppard at University of Bristol, aims to promote the development of new drugs to treat the most common cause of cystic fibrosis, a genetic mutation called F508del.

Dr Sheppard's SRC will investigate the structure of CFTR, which enables chloride ions to cross between cells in the body, to learn how the mutation works, and search for chemicals that repair the faults it causes.

The Cystic Fibrosis Epidemiological Network (CF-EpiNet), led by Professor Di Bilton and her colleagues at Royal Brompton Hospital, will extend the range and use of UK CF Registry data, which contains information for over 99% of the UK CF population, linking it with other health and educational records to help identify treatments that most benefit health, survival and quality of life at critical life stages.

The Trust also announced an open call for applications to establish future SRCs, with successful bids announced in early 2016.

Find out more about the strategic research centre programme at cysticfibrosis.org.uk/src

Tackling the common cause: Dr Sheppard's team is exploring how the F508del mutation works, and possible treatments.

Registry Report

The latest UK CF Registry Data Report presents a snapshot of cystic fibrosis in the UK. This year's figures reveal a rise in transplants for people with CF and the impact of ivacaftor.

10.5%

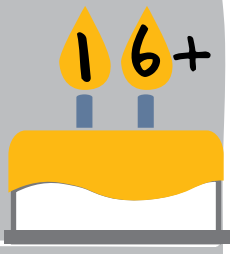
improvement in FEV_1% predicted 6-8 weeks after starting ivacaftor treatment.

72

transplants took place compared to 57 in 2013.

59.3%

of people with cystic fibrosis are aged 16 or older.



70.1%

of adults with cystic fibrosis are in employment or study.

Read the full and 'At a glance' reports at cysticfibrosis.org.uk/registryreports

Could the ancient martial art of tai chi improve the health and wellbeing of people with cystic fibrosis?

Tai chi and you

The Trust has awarded £19,000 to complete research into how combining the centuries-old martial art of tai chi and modern technology could enhance the quality of life for people with cystic fibrosis.

The study, run by London South Bank University in conjunction with Royal Brompton and Harefield NHS Trust, is investigating the impact tai chi has on people with cystic fibrosis as well as the effectiveness of teaching tai chi via Skype and DVD.

The Trust is funding the research as part of an ongoing strategy to invest in projects looking at the impact exercise has on the treatment and management of cystic fibrosis and on the strength of a pilot study which found tai chi had improved the participants' lung function, sleep and anxiety levels.

"The Trust was keen to support the research because not only does it look at the effectiveness of an activity but also the accessibility," Director of Strategic Innovation, Dr Janet Allen, said.

"If this study shows tai chi can be effectively taught using technology, it could open the door to virtual exercise groups which will benefit the CF community as much as it will individuals."

Learn more about tai chi, and find a class near you at www.taichiunion.com.

Sixty Five Roses for Scotland

The Sixty Five Roses Club, an organisation for Trust supporters who pledge to donate more than £1,000 each year, has launched a new branch for Scottish philanthropists.

The branch launched with a reception in Glasgow celebrating the brilliant work being done in cystic fibrosis in Scotland, with speakers including celebrity patron Laura Main, from BBC 1's 'Call the Midwife', Dr Emma France, who is leading a project exploring childhood adherence (see p30), and Allan Gormly, Deputy Chairman of the Trust.

For more information and to sign up visit cysticfibrosis.org.uk/sixtyfive or email Vicky Bratherton, Philanthropy Manager, vicky.bratherton@cysticfibrosis.org.uk.

News in brief

Sixty-Five Roses Ball "a triumph"

The ninth Sixty-Five Roses Ball, held at The Dorchester on Park Lane in June, appropriately raised £65,000 for the Trust.

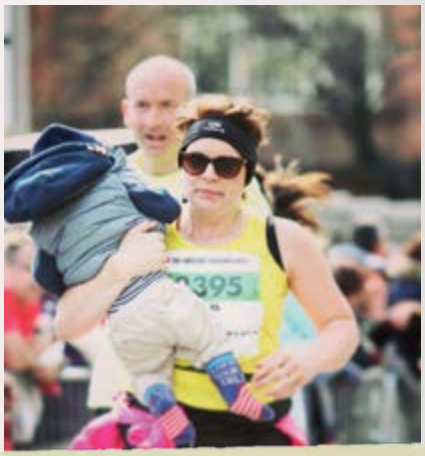
Over 250 people attended the event, which featured an auction run by Channel 4's Raj Bisram and included sporting and film memorabilia, original artwork and VIP concert and travel packages.

Claire Phillips, the Trust's Community Development and Fundraising Manager, said: "The ball's success was testament to the dedication and hard work of the organising committee who epitomise the commitment the CF community has to beating cystic fibrosis for good."

Cystic Fibrosis the power of us

Thank you to all of you who shared your stories and photos with us during CF week to help us harness the #PowerOfUs

Miss Hampshire and Miss Basingstoke got two bobbies on the beat to take part in the strawfie challenge.



Jules crossing the line of the Sheffield half marathon, carrying her son who has CF.

Tom Meredith, one of the physios at Southampton General Hospital, gave the best physio case presentation at the European CF Conference during CF week.

Lovely in yellow: Spinnaker Tower, thanks for lighting up for us.

#PowerOfUs

Rebecca Bow took the plunge for CF Week and throw herself out of a plane.

Annie Hall sent us this great picture of her son James, who despite having cystic fibrosis and things not always being easy, is living this dream.

He has a degree and is a performer with a theatre company in Lincoln. Once again, he will be performing at the Glastonbury Festival this year. Anne and James are grateful to the new drug Kalydeco and the CF team at St James in Leeds, for keeping well.

Parent Pack

Facebook views

10,000+

Parent Pack video received more views during CF Week than many of our long-standing videos.

Many commented on the innovative resource about how it could have helped them when their child was diagnosed.

Eastenders

Combined Facebook and Twitter reach

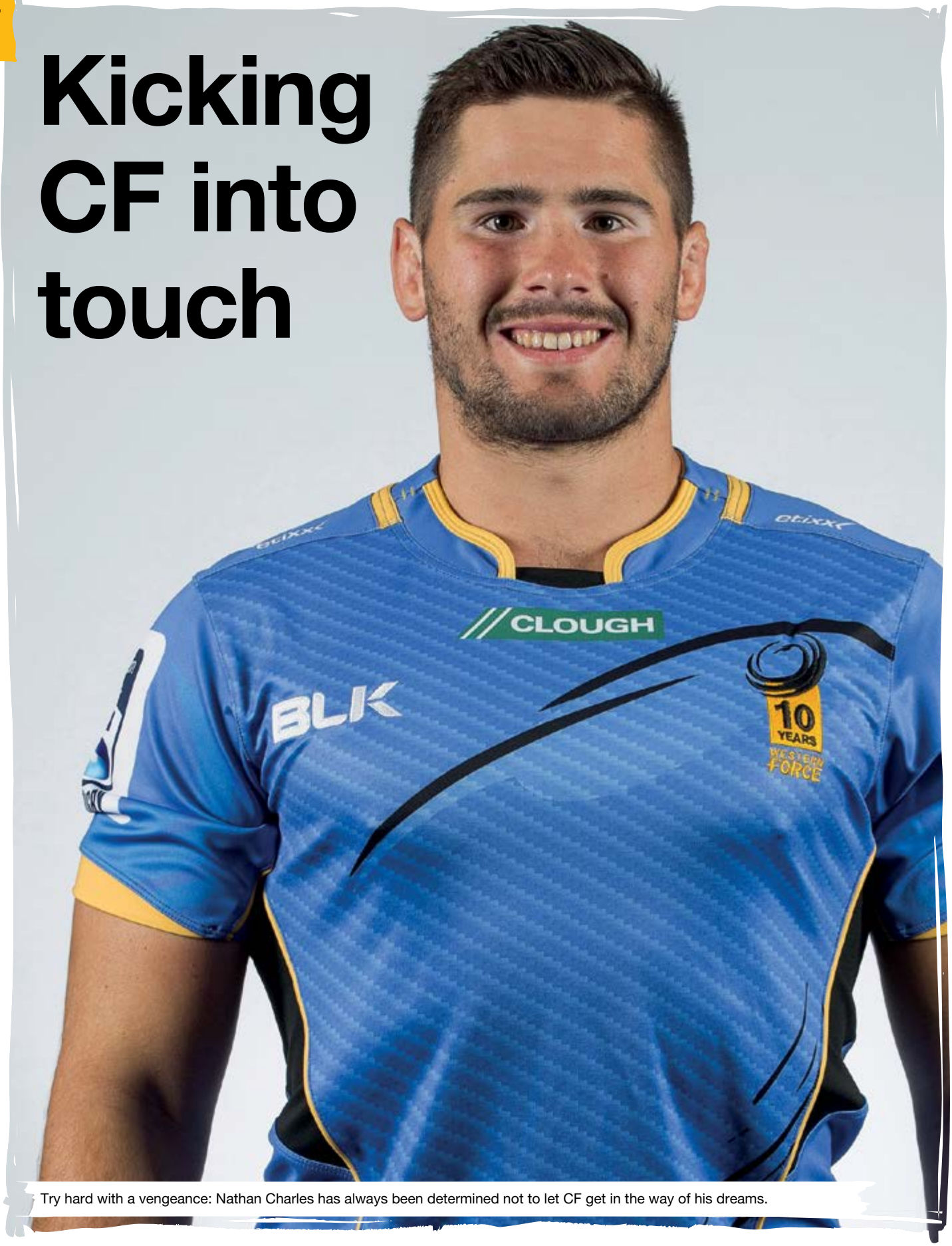
74,000+

Our Eastenders content engaged a new audience and reached beyond the CF community.

Our infographics helped to answer many questions from this new audience.

Don't forget to take a look at our YouTube channel youtube.com/cftrust to view videos from Trust initiatives and events.

Kicking CF into touch



Try hard with a vengeance: Nathan Charles has always been determined not to let CF get in the way of his dreams.

Photo credit: Gordon Pettigrew/ True Spirit Photos

"Ever since I was a young boy I knew rugby was what I wanted to do. I always wanted to be the best I could be."

Photo credit: Gordon Pettigrew/ True Spirit Photos

Playing rugby for your country is the kind of thing schoolboys have dreamed of since the game first began.

On 14 June 2014, a 25-year-old Sydney-born club player stepped onto the pitch at Melbourne's Etihad Stadium to face a fearsome French side, and became one of the few who gets to live the dream. But for Nathan Charles, this was more than just fulfilling the ambitions of a lifelong sports fan. Because when he was diagnosed with cystic fibrosis at birth, Nathan's parents were told he may not live to see his 10th birthday.

Nathan Charles is a star rugby player first and foremost, one who just happens to live with a life-shortening genetic condition. But as the only known professional with CF playing a contact sport, the 26-year-old hooker also embodies the spirit of determination that refuses to accept the limitations imposed by cystic fibrosis.

Now in his fourth year at Perth's Super 15 Rugby team Western Force, Charles has won critical acclaim, and previously played for English premiership side Gloucester Rugby and the Canberra-based ACT Brumbies. So what kind of start in life did he need to give the tenacity and focus to make it to the top? And how did he keep his cystic fibrosis from affecting that?

Charles says he'd like to think his childhood was pretty normal, despite being diagnosed with cystic fibrosis. "As a kid you don't fully understand what the implications are. I remember mum and dad always encouraged me to play as much sport as possible and be active. I remember being on my nebuliser a lot but that was also just a normal part of life. As a child you don't know something is tough or unusual because you don't know any different so I just got on with it."

And get on he did. As a youngster Charles played rugby union and league, basketball, soft ball, tee ball, water polo and soccer.

"As a kid you don't fully understand what the implications are. I remember mum and dad always encouraged me to play as much sport as possible and be active."

"Ever since I was a young boy I knew rugby was what I wanted to do. I always wanted to be the best I could be. I realised it was a possibility when I got invited to join ATC Brumbies in 2008. It was just a case of working hard for the success." ▸

Youthful rebellion, however, came in the form of not always best protecting his health. “Growing up, obviously you sometimes rebel. There would be times, for example when I was staying at a mate’s place that I wouldn’t take my nebuliser because I wanted to be like everyone else.”

It wasn’t until his teens, he admits, that he had any realisation of how serious CF could be. He also realised that exercise could be, as he puts it, “the best medicine.”

“Being active definitely helped my CF because of the way it kept my lungs pumping. I’ve noticed too since that when I am at my fittest, that’s also when I’m at my healthiest.”

Nathan’s attitude obviously contributes massively to how well he’s dealt with the obstacles presented by living with cystic fibrosis. “I wouldn’t say I’ve had obstacles so much as challenges. For example, when travelling for work all the time it can be difficult to carry all the right medication with you. You have to be disciplined to do your nebuliser morning and night, even when you’re on tour with the guys.”

But it’s clear that Charles has surrounded himself with the right kind of support. “As I was growing up, my parents were just so encouraging. They would pick me up, drop me off, watch me play games. They never let anything get in the way of me competing.” That, in turn, inspired Charles to ensure nothing got in his way either.

“I’ve never been surrounded by pessimists that think you can’t achieve. And that sort of foundation definitely inspired me.” Growing up, Charles felt blessed to be surrounded by “a lot of great family members and friends that guided me along the way.”

“Being active definitely helped my CF because of the way it kept my lungs pumping. I've noticed too since that when I am at my fittest, that's also when I'm at my healthiest.”

A Force to be reckoned with: Nathan has had a starring role as hooker for Perth’s Super 15 rugby team Western Force.

Photo credit: Gordon Pettigrew/ True Spirit Photos

"I am the only professional rugby player in the world that has CF; I'd like to take that and inspire people and let them know they can achieve too."

Photo credit: Gordon Pettigrew/ True Spirit Photos

Nowadays, he can count his long-term girlfriend as another valuable cheerleader. "She is just so amazingly supportive." He doesn't know what the future holds, but he knows having children would present "a new challenge to my health that I've not had to deal with yet."

"Sometimes I will get a bit ill... I have learnt that the way to deal with it is to be proactive rather than reactive."

Although Charles wouldn't directly credit rugby over another sport, he believes it has definitely helped his cystic fibrosis. "There isn't one sport that's better than another but I do think all the running contributes massively. Of course that doesn't mean I don't take any medication. I still have to take a load of tablets and have my nebuliser to hand. Thankfully they are smaller these days."

Still, despite his fitness, Charles has battled through sickness. "Sometimes I will get a bit ill, a chest infection or bug and that slows me down. I have learnt that the way to deal with it is to be proactive rather than reactive. That means the minute I feel just a little bit under the weather I jump on it at that stage, get to the doctors and make sure it doesn't get any worse. And then of course you just have to push on through."

While Charles won't yet be playing in the World Cup, it remains an ambition for him, as well as making a finals squad with the Western Force team. And he has non-sporting ambitions too. With only six to eight years left to play at professional level, he's currently training to be a mortgage broker and making plans to travel extensively. And then there's his motivational speaking career.

"I am the only professional rugby player in the world that has CF; I'd like to take that and inspire people and let them know they can achieve too."

So what is his advice for anyone wishing to pursue a sporting career, or indeed anyone with CF wishing to live a long and healthy life?

"Try and be as fit as you can possibly be. Understand your body and what kind of bugs you might get. And remember that exercise is the best medicine."

Getting active

Susan uses the Forth

165 feet doesn't seem a long way until you're going backwards down Edinburgh's Forth Rail Bridge holding onto a rope, but when Susan Murray saw her 76-year-old mother racing down the iconic crossing, her fears abated and she began her descent. Susan 'went over the top' in memory of her husband Robert.

"Robert and I met through mutual friends," Susan recalls. "We didn't date long and were married for five years. He was heavily into art and painting."

"I'd never abseiled before but being surrounded by my team made it so much easier."

"I decided to mark what would have been his 50th birthday year with an abseil to raise money for the Cystic Fibrosis Trust. Robert was on the transplant list and twice we were told there were organs available, but both times were fruitless. The Trust helped us with the cost of travelling from Scotland down to Newcastle for Robert's assessment and provided fantastic support throughout. They helped us navigate a mountain of paperwork."

Susan and her team of friends and family raised over £5,500. "When you tell someone a personal story, I think they are more inclined to sponsor you than they otherwise would be," Susan says.

"The abseil itself was a bit nerve-racking, especially because they'd had to cancel abseiling the day before because it was too windy. I'd never abseiled before but being surrounded by my team made it so much easier."

"I'd love to do more for the Trust in the future. This was such a great way to celebrate Robert's life."

Wish you were here? Team Murray celebrates after abseiling Scotland's most famous bridge.

Be inspired by Susan, and take on a challenge to raise money for the Trust – visit cysticfibrosis.org.uk/events.

Don't forget you can find highlights of the latest news and read comments on our facebook page.'Like' our page and stay connected with the Trust at facebook.com/cftrust.

cysticfibrosis.org.uk/brightideas

Cystic Fibrosis not going to stop me

Helen Barrett Bright Ideas Awards

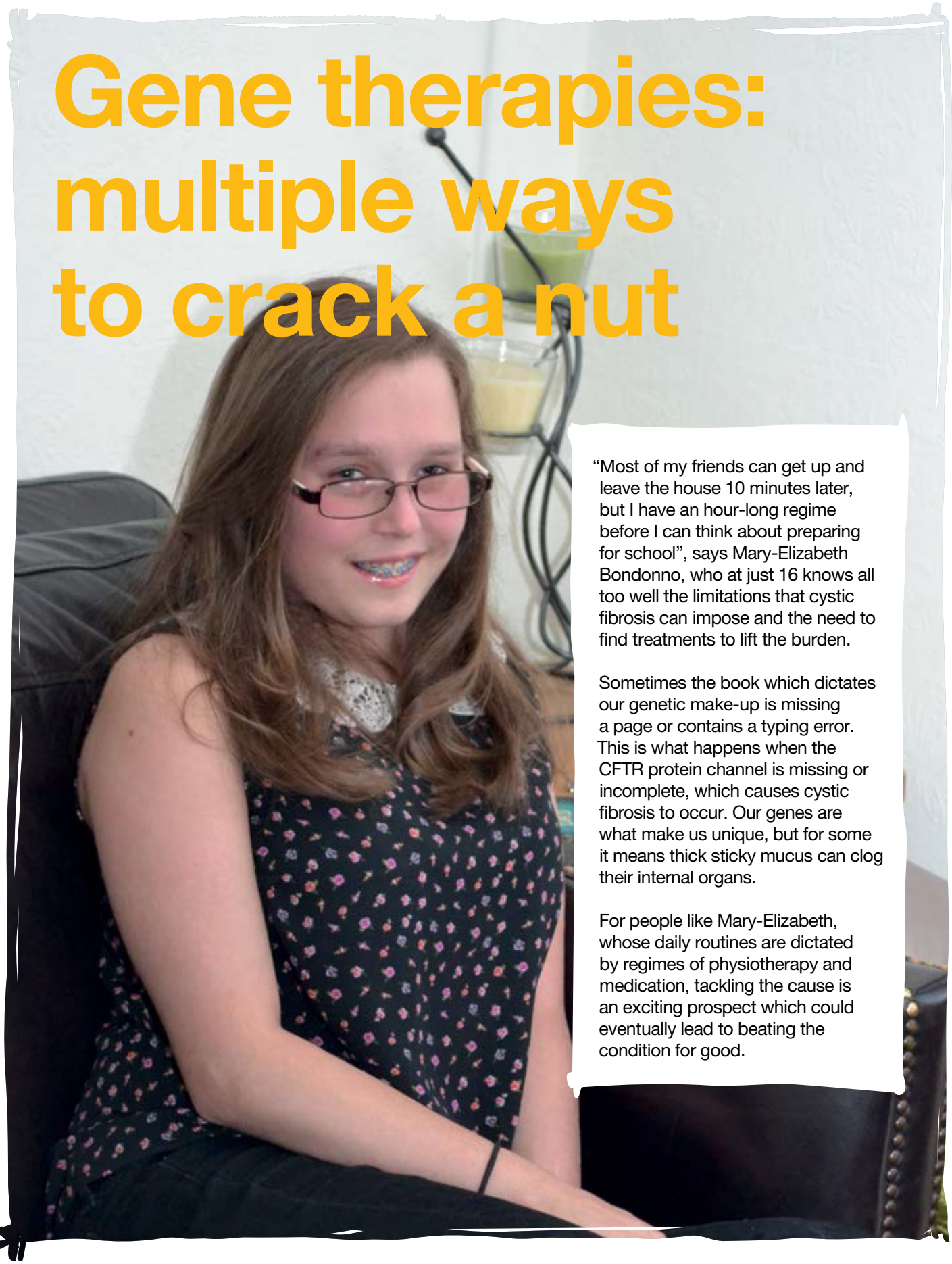
Live the life you want by turning your passion into £££s!

Whatever you need to kick-start your dream career, if you're aged 16–35 you can apply for a grant of up to £1,000 and expert mentoring to help you on your way.

Helen Barrett dreamed of setting up her own business, and she didn't let cystic fibrosis hold her back. Her gym is thriving, and through these awards set up in her memory we hope to inspire others to make the leap too.

Go to cysticfibrosis.org.uk/brightideas now!

Gene therapies: multiple ways to crack a nut



"Most of my friends can get up and leave the house 10 minutes later, but I have an hour-long regime before I can think about preparing for school", says Mary-Elizabeth Bondonno, who at just 16 knows all too well the limitations that cystic fibrosis can impose and the need to find treatments to lift the burden.

Sometimes the book which dictates our genetic make-up is missing a page or contains a typing error. This is what happens when the CFTR protein channel is missing or incomplete, which causes cystic fibrosis to occur. Our genes are what make us unique, but for some it means thick sticky mucus can clog their internal organs.

For people like Mary-Elizabeth, whose daily routines are dictated by regimes of physiotherapy and medication, tackling the cause is an exciting prospect which could eventually lead to beating the condition for good.

Getting involved: Mary-Elizabeth Bondonno was proud to take part in the gene therapy trial.

Gene therapies target the root cause of CF, the faulty CFTR gene. There are two different ways that gene therapy can be used: one inserts a normal extra copy of the gene into lung cells to compensate for the defective gene and the other corrects the mutation in the gene itself.

Inserting the normal CFTR gene

The Trust has, for many years, invested in the Gene Therapy Consortium (GTC), which is investigating using a nebuliser to deliver a normal extra copy of the gene into the lungs. This area hit the headlines with results of the latest clinical trial, which Mary-Elizabeth took part in.

Professor Eric Alton, who led the trial, says: "The aim of gene therapy has always been prevention, but the key issue at the moment is the delivery of the gene into the cell. Progress with the trial is as expected, we know that it is not a cure at this stage, but we are hopeful."

Although modest and variable, the trial demonstrated promising results, which prove that repeated dosage of gene therapy can produce some benefit. Mary-Elizabeth explains: "Although I still had to take my medication on the trial, my lung function went up and I felt that I had much better concentration on my school work and I was able to run around at the park with my brothers without getting too out of breath – something I couldn't do before."

The phase 2b study delivered the gene using fatty droplets or liposome (wave 1) and the GTC is also exploring the use of viral delivery of the CFTR gene (wave 2). The Trust recently announced £500,000 of further funding for the consortium, which aims to insert the gene encoding the CFTR in the airway cells.

"My hope is that one day genetic therapies will give me my life back – no longer having to get up hours earlier each day to do physio, or rattle around with all the medication."

Gene Correction

The second generation of gene therapies focus on correcting the gene mutation inside the cell. This is called editing and can either be at the gene or RNA (the genetic messenger for protein production) level. This work is all at an early stage and has yet to progress to clinical trials. However, the Trust is investing in two different exploratory projects in the area of gene editing, one led by Dr Harrison, and one by Professor Floto and Dr Vallier.

Dr Harrison and his team are looking to locate and cut out the faulty sequence in the gene using ▸

so-called ‘molecular scissors’ and then to introduce the correct sequence. This has been achieved in cells in the test tube.

Dr Vallier and Professor Floto are using stem cells as opposed to adult cells to make corrections in the CF gene. One type of stem cell (IPSCs) can be derived from any individual. These IPSCs have the unique ability to divide into numerous types of body cell. The project is exploring gene editing in IPSCs derived from people with CF caused by a ‘stop’ codon, a class of mutation that stops the CFTR protein being produced. The aim is to correct the stop codon in the IPSCs, and ultimately to work out how these cells can be used to treat that individual. The first part of this has been achieved. The second part is more tricky but without investment in research at this stage, we will never know the answer.

> “...approaches using gene therapies are beginning to yield promising results both in the laboratory and in clinical studies.”
>
> - Dr Janet Allen

The Trust is also contributing to a study, co-funded with Action Medical Research, investigating a further approach to gene therapy that does not involve the CFTR gene directly. This project aims to switch off a gene called ‘ENaC’. If successful, this should help protect the lungs from dehydration and help clear the mucus by making it less sticky.

The Cystic Fibrosis Trust brought together experts from a range of disciplines at a round table meeting in June to explore the ways genetics and gene therapies can be used to treat cystic fibrosis. The group, which included Bill Skach and Preston Campbell from the Cystic Fibrosis Foundation in the States, discussed the future of gene therapies for cystic fibrosis.

Dr Janet Allen, Director of Strategic Innovation, says: “Twenty-five years after the gene responsible for cystic fibrosis was discovered, approaches using gene therapies are beginning to yield promising results both in the laboratory and in clinical studies. Various approaches are being tested and the Trust needs to stay at the cutting-edge of this science to ensure all options are explored. The visit by our colleagues from the Cystic Fibrosis Foundation provided a unique opportunity to bring together the key UK scientists and clinicians working in this area and explore ways that UK scientists can join forces with those in the USA to find the best treatments as quickly as possible.”

Mary-Elizabeth sums up the real importance of investing in gene therapies in her hopes for the future: “My hope is that one day genetic therapies will give me my life back – no longer having to get up hours earlier each day to do physio, or rattle around with all the medication I’m taking. Free to enjoy myself and not worry about my schedule.”

Find out more about the Trust’s ground-breaking research programme at cysticfibrosis.org.uk/research.

Day in the life

Tara Illingworth, 28, is the editor and curator of 'The Brew', a lifestyle website she launched in 2012 after moving to Manchester from Florida.

"Cystic fibrosis doesn't have to win. I believe we can live happy lives, pursuing our passions and dreams." – Tara

When faced with a lifetime sentence like an incurable disease, it's easy to let it overwhelm you and affect everything you do, but the beautiful thing is we each have a choice. When I was diagnosed with cystic fibrosis at six months, my parents chose to fight for a cure and not let the sentence rule my life. Growing up taking medication and filling my mornings and evenings with breathing treatments, I've tried to live by that same example. Cystic fibrosis doesn't have to win. I believe we can live happy lives, pursuing our passions and dreams.

Cystic fibrosis caused me to develop cirrhosis of the liver, which meant I needed liver and pancreas transplants. Two years later, I received my new organs, which was a tough and uncertain experience. My biggest piece of advice for anyone awaiting a transplant is to be diligent about staying as healthy as possible until that organ arrives. What kept me most positive was surrounding myself with an amazing support group of family and friends who continue to make a world of difference. Today that support exists largely from my husband who, it turns out, is a carrier of the CF gene. As a result, the focus of our energy right now is Pre-Implantation Genetic Diagnosis (PGD) to ensure, after many years of being told I couldn't have children, we can have a beautiful child that we can (almost) guarantee won't have cystic fibrosis.

I started The Brew to document our journey, along with life abroad, travel, fashion and the struggles of everyday life. It has grown into a commercial platform with up to 12,000 monthly readers and has given me the opportunity to leave my full-time PR job. The tricky part is knowing when to post about the realness of CF in my life and when to hold back – it's a balancing act. I'm also looking forward to spending time on the other projects close to my heart, like 'Rock n' Roses', a CF fundraiser I started in Florida that raised over $35,000 in two years, which I'm hoping to resurrect on this side of the pond soon.

My typical day begins with a few rounds of airway clearance, autogenic drainage (AD), followed by two breathing treatments. I get in at least 15 minutes of exercise every morning. After that, I eat breakfast, take my morning medicine and tackle my job as an editor. Some days consist of developing content and on others admin takes priority, with lunch squeezed in. At night I do a few more rounds of AD and conclude with three additional breathing treatments and evening medication.

The biggest obstacle I face is ensuring life doesn't get so hectic that I neglect my treatments, especially when we are busy traveling. Planning is everything and I know prioritising my health will pay off in the long run. The most important thing to remember is that CF doesn't need to define you.

Check out Tara's lifestyle website at www.TheBrew.me

Activity Unlimited

To borrow a phrase, it's not the winning that matters, it's the being active that counts.

October 2012 and Dr Jonathan King is on a ventilator being rushed from Heathrow to hospital in the back of an ambulance.

Jon had moved to Mumbai four months earlier to work as a doctor, despite being warned of the danger the pollution and climate posed to his health. Being told he couldn't do something wasn't foreign to him – after all he'd been told he couldn't handle the rigours of studying medicine and he'd proved them wrong.

Five months passed and Jon's condition deteriorated; he could neither feed nor dress himself. "They told me I needed a lung transplant," Jon recalls. "I wanted to hold out for as long as possible, this was the final roll of the dice. My nurse told me '12 months is a long time'. I'll never remember anything as clearly as I remember that moment."

Jon left hospital in April 2013 no longer requiring oxygen constantly, but needing it to walk anywhere and sleep. “I decided I’d exercise in the vague hope it would improve my quality of life before the transplant. I thought there was an outside chance it may even delay it for a few months.” Jon and his oxygen hit the gym.

Jon’s day of reckoning was 17 July 2013. His exercise regime, which began with oxygen-assisted 10-yard walks had progressed to 4km oxygen-free runs, but Jon had returned to hospital for his assessment. “I didn’t think I’d need the transplant anymore and the surgeons agreed. I’d exercised my way out of the dead end.”

“I decided I'd exercise in the vague hope it would improve my quality of life before the transplant.” - Jon

Feeling it: Michelle Lincoln says that the benefit of yoga twice a week really tells.

On the road: Dr King set off from Richmond on a 1,200-mile ride to Barcelona to raise awareness about the importance of being active.

The benefits of exercise for managing cystic fibrosis have been reported for two decades. Physical activity improves lung function, which helps fight infection and reduces the need for medication and transplants, which is why the Trust is developing ‘Activity Unlimited’, a project aimed at getting the entire cystic fibrosis community active.

“Jon is the embodiment of why being active matters and the value of healthy lungs,” says Paul Rymer, Head of Principal Involvement. “The breadth of Activity Unlimited is what makes it exciting. Physical activity is critical in the fight against cystic fibrosis, but we aren’t just interested in getting teenagers playing football on the weekends, we want people of all ages doing anything to get their lungs working.”

“We want parents playing with their kids on the trampoline, students playing sport in the schoolyard, people of all ages running, dancing, and swimming. We want people doing Pilates or boxing or using game systems like Wii and Kinect.”

“I knew I needed to improve my physical health, having been diagnosed with CF in my 30s.” - Michelle

Michelle Lincoln loved dancing and running in senior school but after graduating, she confesses, most of her exercise was slightly different. “The only exercise I did was dancing in nightclubs. I knew I needed to improve my physical health, having been diagnosed with CF in my 30s, and I’d tried aerobics and the gym but due to my lungs, I needed something less cardio heavy.” ▸

Fighting fit: Aaron Aby swapped the football pitch for the MMA ring.

Michelle does two hours of yoga a week and says the impact is palpable. "I do lots of walking and can now walk faster for longer thanks to yoga. It also helps my sleep and I really notice the difference missing a session makes."

"Exercise is crucial and achievable for everyone, but particularly those with CF." - Jon

The Trust has established an Activity Unlimited work group comprising people with cystic fibrosis, fitness experts, clinicians and Trust staff to develop the programme. "The group is integral to ensuring we can enable and inspire people to be more active. We've looked for diversity and expertise in the members, but we want contributions from everyone," Paul says. "The more contributors we have, the stronger the project will be."

"The work group is the first stage of a wider plan. Our next step is to engage sporting ambassadors to spread the Activity Unlimited message and then develop corporate partnerships to offset the cost of participation, which can be prohibitive. Whether that's offering discounted equipment, classes or gym memberships, we want to make sure exercise is accessible. We already offer grants for people to buy things like trampolines for their children and we want to help more of the community get active."

One of the sporting ambassadors is Aaron Aby, a professional mixed martial arts fighter who played under-age football for Wales alongside Gareth Bale, but was released by a professional football club for having cystic fibrosis. Aaron, 25, has raised over £20,000 for the Trust and often speaks at schools, dinners and charity events. "Aaron's passion is what we want to instil in our community," Paul says. "He is committed to sharing his story and to showing people what he can do. His short film 'Fighting to Breathe' is inspirational."

Integrating an exercise programme into an increasingly digital world is a major challenge, Paul says. "We are investigating how to harness technology to benefit the programme and participants. We are looking at an app to help people manage their activity. The Activity Unlimited group uses Skype to overcome cross-infection issues."

"We want people to give one another the confidence to try something new, which is why the Trust website, social media and (online collaboration space) 'Hack It Up' are so critical to the project. We want the major source of inspiration to be our community."

On 30 July 2015, Jon King arrived in Barcelona 13 days after he set off from London on a 1,200-mile fundraising ride. "I feel I've reinforced my main messages," Jon says. "Firstly that exercise is crucial and achievable for everyone, but particularly those with CF, and secondly, the CF community, in fact the chronic disease community as a whole, should never feel we shouldn't have the same goals that others have."

What does being active mean to you?

Your experiences will inform our Activity Unlimited programme – we want to know about your challenges and triumphs in the quest to stay healthy. Which sports or activities fit in with your lifestyle, and how do you make exercise work for you? Share your stories and advice online with #ActivityUnlimited to inspire others to get active and stay active.

Out in the community

Grace Paget (L) and Claire Philips (R) meet The Streets singer Mike Skinner at the Highgate School Charity Summer Fair.

In the last two years, hundreds of schools across the UK have raised a fantastic total of £160,000 for the Trust, but for Community Development and Fundraising Manager, Gemma Walder, school fundraising activities are about more than just money.

"We get lots of schools doing Big Bounces, sponsored walks and cake bakes, but joining in and raising awareness are just as important as the amount raised," Gemma says. "Next year we will launch our new schools' fundraising pack filled with ideas, posters and guides to organising and running an event on behalf of the Trust."

"We rely on the generosity and kindness of the public so it is important we support them as much as possible when they are going to the effort of supporting our fight against cystic fibrosis."

Highgate School chose the Trust to be one of its three charity partners for 2015 and celebrity parents past and present, including Coldplay's Chris Martin and Mike Skinner from The Streets, were on hand to celebrate the school's 450th birthday at the Charity Summer Fair. The school also published a cookbook filled with hearty recipes from parents and illustrations from students.

The event and book raised over £10,000, and Claire Phillips and Grace Paget from the Trust ran a busy stall handing out leaflets and information about cystic fibrosis and the Trust's work.

Over in Northern Ireland, a group of Year 13 business studies students at Sacred Heart College, Omagh, raised £800 through a half marathon, 5km run and egg hunt around the school while students at Rathmore Primary in Bangor held a fitness day which raised over £6,000.

Every penny can make a difference. Enterprising eight-year-old Isabelle Smith, of Tidmarsh, Berkshire, organised a 'guess the number of lollies' competition, with some help from her mum, in honour of her friend Tilly. Isabelle charged her classmates to guess and raised £46, which her dad doubled.

"Isabelle's story is a great example of how simple fundraising can be and this is what we hope to illustrate with the new schools' fundraising pack," Gemma explains. "Not every event needs to be on a massive scale but they do need to be about having fun, getting people involved, raising awareness and helping the Trust beat cystic fibrosis."

If you and your school would like to get involved in fundraising go to cysticfibrosis.org.uk/schoolfun. You can get inspired by our 'A to Z of Fundraising Ideas' and get loads more information about how we can support and promote your events!

Two-point perspective

Juliette describes what it was like when she and her husband Mark discovered that their son Sammy had cystic fibrosis, and CF Nurse Alison provides a professional view on new diagnosis.

Mark, my husband, and I were delighted when Sammy arrived in the summer of 2012. He was, and still is, beautiful. His brother Joseph was three at the time, and was very proud to be a big brother and interested to see what all the fuss was about. We came home in the usual way after the birth, exciting, overwhelmed and a little tired!

Sammy seemed fine for his first week, I was breastfeeding a lot but then you expect to in the early days. But then he started to lose weight and wouldn't settle; initially I thought he had reflux. It was really hard because I was feeding Sammy constantly and yet he still appeared to be hungry and often inconsolable. As time went on, I thought something was wrong. Joseph was at nursery and so the normal day-to-day routine had to continue. I would put both boys in the car to go to nursery and often Sammy would cry frantically all the way there until I could feed him again. It was exhausting physically and emotionally.

When Sammy was seven weeks old, I received a call from the health visitor to say that they had test results from the Guthrie test. I didn't know what the Guthrie test was and didn't want to find out, I was really frightened. She asked if I had someone with me, so I phoned Mark and he came home. It was several hours until the specialist nurse arrived. This was a terrible few hours. We didn't know what to think and I feared the worst. I thought my little boy was going to die in infancy or be disabled.

> "I asked her if Sammy would die young, because obviously this was my greatest fear."

When Alison, our CF Nurse, arrived she told us gently but clearly that she thought Sammy had cystic fibrosis. She said he needed to have a sweat test, which she booked for us, but that CF was likely to be diagnosed. She answered all our questions, sensitively but honestly and we both felt well supported. I asked her if Sammy would die young, because obviously this was my greatest fear, and she was very reassuring. In a way, because I had those few hours to think the worst, the diagnosis of CF was, initially, a strange relief.

Sammy's health steadily improved after his diagnosis was confirmed and treatment began. He fed well, put on weight and began to sleep better too!

Alison Betteridge
CF Nurse
Norfolk and Norwich University Hospital

I have worked as a CF Nurse for 21 years, but I cannot say that breaking the news has become any easier. However I have learned much from parents about how significant that day is. They can always remember who broke the news and their initial thoughts and fears.

Most parents know little about the condition and have great fears about what it means. We try to address their greatest fears as soon as possible – will my child die soon and will they have disabilities? We ensure that they get the message early on that with early treatment and additional care we expect their child to do well, go to normal schools and grow into adulthood. We will talk about children we have seen who have been cross-country runners, hold down good jobs and now come to see us with their own children.

We try to keep the time from parent knowing something is wrong to getting the diagnosis to a minimum. We talk to them via the telephone directly after the health visitor has given them the 'CF is suspected' leaflet, and arrange to meet them at home the next morning. This gives them the opportunity to express any concerns in their own familiar environment and we can undertake sweat testing there too.

We then return to the hospital and arrange to meet them at lunchtime with the results of the test and for them to meet the CF centre consultant. Together with the consultant we talk through the results and demonstrate how to give medications. We aim to equip parents with the skills they need and the explanations to help other family members understand.

In my experience, even if children have relatively few symptoms, parents are alerted to something not being quite right and are relieved when we can provide an explanation.

Most parents know little about the condition and have great fears about what it means.

We also use Cystic Fibrosis Trust resources to help parents understand why their emotions may seem to be 'all over the place'.

They may have chosen to become parents but no parents would choose to become a carer; however people's strength in coping never ceases to amaze me.

Juliette and Sammy feature in our new 'Parent information pack', which offers reassurance and support for families coming to terms with a new diagnosis of cystic fibrosis.

Find out more and watch our film at cysticfibrosis.org.uk/newdiagnosis.

Don't forget you can find highlights of the latest news on our Twitter feed. Stay connected with the Trust at twitter.com/cftrust. **Follow us on Twitter @cftrust.**

In the lab

Dr Emma France from the Nursing Midwifery and Allied Health Professions Research Unit at the University of Stirling talks about the pioneering research she is leading, using multimedia to help families and carers with children's home physiotherapy.

What is the project you are working on? What does it involve?

For the last year I have been leading the 'SCooP' (Supporting Children's Physiotherapy for cystic fibrosis) project to develop a short film in partnership with a group of parents of young children aged eight and under with cystic fibrosis. The film is to encourage families and carers to stick to their child's home chest physiotherapy routine to help them to stay well. It features real families talking about their experiences of CF and chest physiotherapy. For the next part of the project we are currently looking for more families who have a child with CF to watch the film to see if it helps them.

What inspired you to go into this profession?

My background is in psychology (but don't hold that against me please!), which I chose to study because I am fascinated in understanding why people think and behave in the ways they do. I started off in telecoms research, but I was uncomfortable with work that was focused on trying to get people to buy more technological gadgets, so I looked for an opportunity to work in a non-commercial research role, where I might be able to help people! Now I am a senior lecturer and my research focuses on families and the impacts of long-term health conditions.

What excites/interests you about the work that you are involved in?

The potential to make a difference to the lives of families affected by illness is what excites me about my work. It's been great working in partnership with families to create the film as part of the SCooP project and seeing the final film coming together.

Why did you choose the field of cystic fibrosis? How do you find it?

I am interested in how families are affected by serious health conditions and by how family life can affect how adults and children manage their health conditions. I chose CF because it is one of the most common genetic diseases in the UK, the treatment for which has a considerable impact on those with CF and their families.

Tell us something non-science related about yourself!

When I'm not working I like to do yoga, dance, go out in the Scottish hills walking and spend time with my family. I am also a qualified scuba diver but I have to admit to being a bit scared of large fish!

Don't forget to take a look at our YouTube channel youtube.com/cftrust to view videos from Trust initiatives and events.

genotypematters.org

Cystic Fibrosis as individual as you are

Get the treatment that's the right fit for you.

When it comes to cystic fibrosis treatment, there's no 'one size fits all'. As we move into the era of personalised medicine, over the coming years we will be seeing more treatments that are tailored to you.

But in order to access these personalised medicines, you need to know your genotype.

To discover yours, talk to the staff at your CF centre at your next visit. Knowing your genotype may mean you can access new drugs that could transform your quality of life.

Your genotype could already be on record. But if it isn't, then the Cystic Fibrosis Trust will cover the costs of a simple test to reveal yours.

Visit genotypematters.org

Know your genotype.

#genotypematters

ONE SIZE FITS ALL

19 25

THE ORIGINAL
CHAMPNEYS
HEALTH SPA

STAY WITH CHAMPNEYS

FROM £199 PER PERSON

In support of the Cystic Fibrosis Trust Champneys are proud to offer a fantastic stay at any one of our four health spas for a heavily discounted rate for anyone living with cystic fibrosis in the UK and a guest or parents/carers of anyone under 18 living with cystic fibrosis. From £199 per person you can enjoy a two night stay, delicious meals, full use of our spa and fitness facilities and one 25-minute treatment (massage or facial).

FROM £199 PER PERSON AT CHAMPNEYS HENLOW (BEDFORDSHIRE) AND CHAMPNEYS SPRINGS (LEICESTERSHIRE) OR £219 PER PERSON AT CHAMPNEYS FOREST MERE (HAMPSHIRE) AND CHAMPNEYS TRING (HERTFORDSHIRE)

TO BOOK EMAIL SUPERVISORS@CHAMPNEYS.COM QUOTING 'CYSTIC FIBROSIS TRUST OFFER'

REFRESH. RESTORE. REWARD.